



## DCAF17 gene

DDB1 and CUL4 associated factor 17

### Normal Function

The *DCAF17* gene provides instructions for making a protein whose function is unknown. The gene is active (expressed) in several organs and tissues in the body, including the brain, skin, and liver. Within cells, the protein produced from this gene is found in the nucleolus, which is a small region inside the nucleus where cell structures called ribosomes are assembled. It is not clear whether the DCAF17 protein plays a role in this process.

### Health Conditions Related to Genetic Changes

#### Woodhouse-Sakati syndrome

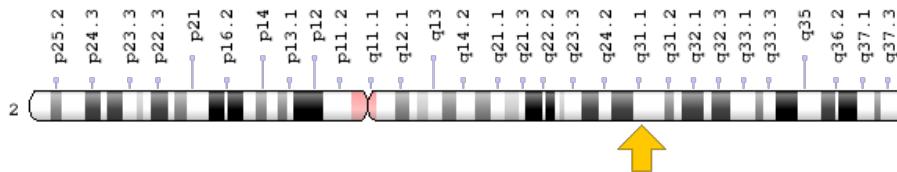
At least 11 *DCAF17* gene mutations have been identified in people with Woodhouse-Sakati syndrome, a disorder that affects the body's network of hormone-producing glands (the endocrine system) and the nervous system. This condition is characterized by delayed or absent puberty, hair loss, abnormal movements, hearing loss, and intellectual disability.

Most of the *DCAF17* gene mutations that cause Woodhouse-Sakati syndrome result in a protein that is abnormally short and breaks down quickly or whose usual function is impaired. Loss of DCAF17 protein function likely accounts for the features of Woodhouse-Sakati syndrome, although it is unclear how a shortage of this protein leads to hormone abnormalities and the other signs and symptoms of this condition.

## Chromosomal Location

Cytogenetic Location: 2q31.1, which is the long (q) arm of chromosome 2 at position 31.1

Molecular Location: base pairs 171,434,166 to 171,491,029 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- C2orf37
- FLJ13096

## Additional Information & Resources

### Educational Resources

- The Cell: A Molecular Approach (second edition, 2000): The Nucleolus  
<https://www.ncbi.nlm.nih.gov/books/NBK9939/>

### GeneReviews

- Woodhouse-Sakati Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK378974>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DCAF17%5BTIAB%5D%29+OR+%28DDB1+and+CUL4+associated+factor+17%5BTIAB%5D%29%29+OR+%28C2orf37%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+OR+%28Woodhouse-Sakati%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

### OMIM

- DDB1- AND CUL4-ASSOCIATED FACTOR 17  
<http://omim.org/entry/612515>

## **Research Resources**

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=DCAF1%5Bgene%5D>
- HGNC Gene Family: DDB1 and CUL4 associated factors  
<http://www.genenames.org/cgi-bin/genefamilies/set/498>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=25784](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=25784)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/80067>
- UniProt  
<http://www.uniprot.org/uniprot/Q5H9S7>

## **Sources for This Summary**

- Agopiantz M, Corbonnois P, Sorlin A, Bonnet C, Klein M, Hubert N, Pascal-Vigneron V, Jonveaux P, Cuny T, Leheup B, Weryha G. Endocrine disorders in Woodhouse-Sakati syndrome: a systematic review of the literature. *J Endocrinol Invest.* 2014 Jan;37(1):1-7. doi: 10.1007/s40618-013-0001-5. Epub 2014 Jan 8. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/24464444>
- Alazami AM, Al-Saif A, Al-Semari A, Bohlega S, Zlitni S, Alzahrani F, Bavi P, Kaya N, Colak D, Khalak H, Baltus A, Peterlin B, Danda S, Bhatia KP, Schneider SA, Sakati N, Walsh CA, Al-Mohanna F, Meyer B, Alkuraya FS. Mutations in C2orf37, encoding a nucleolar protein, cause hypogonadism, alopecia, diabetes mellitus, mental retardation, and extrapyramidal syndrome. *Am J Hum Genet.* 2008 Dec;83(6):684-91. doi: 10.1016/j.ajhg.2008.10.018. Epub 2008 Nov 20.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/19026396>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2668059/>
- Alazami AM, Schneider SA, Bonneau D, Pasquier L, Carecchio M, Kojovic M, Steindl K, de Kerdanet M, Nezarati MM, Bhatia KP, Degos B, Goh E, Alkuraya FS. C2orf37 mutational spectrum in Woodhouse-Sakati syndrome patients. *Clin Genet.* 2010 Dec;78(6):585-90. doi: 10.1111/j.1399-0004.2010.01441.x.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20507343>
- OMIM: DDB1- AND CUL4-ASSOCIATED FACTOR 17  
<http://omim.org/entry/612515>
- GeneReview: Woodhouse-Sakati Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK378974>

- Hdiji O, Turki E, Bouzidi N, Bouchhima I, Damak M, Bohlega S, Mhiri C. Woodhouse-Sakati Syndrome: Report of the First Tunisian Family with the C2orf37 Gene Mutation. *J Mov Disord.* 2016 May;9(2):120-3. doi: 10.14802/jmd.16003. Epub 2016 May 25.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/27240811>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4886203/>
- Steindl K, Alazami AM, Bhatia KP, Wuerfel JT, Petersen D, Cartolari R, Neri G, Klein C, Mongiardo B, Alkuraya FS, Schneider SA. A novel C2orf37 mutation causes the first Italian cases of Woodhouse Sakati syndrome. *Clin Genet.* 2010 Dec;78(6):594-7. doi: 10.1111/j.1399-0004.2010.01447.x.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/21044051>

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